

# Leukaemia Section

## Review

### **t(10;14)(q24;q11) TLX1/TRD**

### **t(7;10)(q34;q24) TRB/HOX11**

**Tatiana Gindina, Karolien Beel**

Cytogenetics Lab, Raisa Gorbacheva Memorial Institute of Children's Oncology, Hematology and Transplantation at First Pavlov St. Petersburg State Medical University, Saint-Petersburg, Russia; [tatgindina@gmail.com](mailto:tatgindina@gmail.com) (TG); Center for Human Genetics, UZ Leuven, Belgium; [karolien.beel@uzleuven.be](mailto:karolien.beel@uzleuven.be) (KB)

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## Abstract

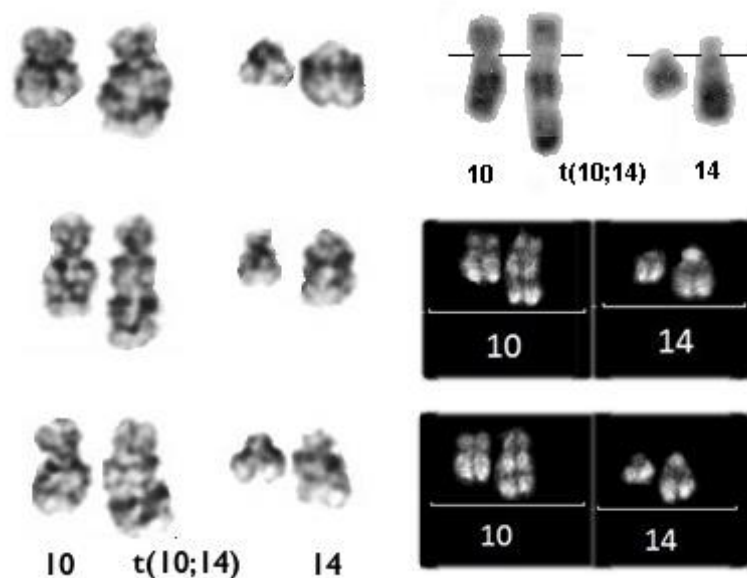
Review on t(10;14)(q24;q11) and t(7;10)(q34;q24) translocations, with data on clinics, and the genes involved.

### KEYWORDS

chromosome 10; chromosome 14;  
t(10;14)(q24;q11); t(7;10)(q34;q11); TLX1; TRD;  
TRB; T-cell Acute lymphoblastic leukemia

## Identity

t(10;14)(q24;q11) or t(7;10) TLX1/TRD or TRB



Left: Partial karyograms showing t(10;14)(q24;q11), GTG-banding - Courtesy Tatiana Gindina. Right: top: t(10;14)(q24;q11) R-banding - Courtesy Pascale Cornillet-Lefebvre and Stéphanie Struski; middle and bottom: t(10;14)(q24;q11) R-banding - Courtesy Karolien Beel, Peter Meeus and Lucienne Michaux

## Clinics and pathology

### Disease

T-cell acute lymphoblastic leukemia (ALL) and non-Hodgkin lymphoma (NHL) with medullary involvement.

A t(10;14)(q24;q11)TLX1/TRD was found in 69 cases of T-ALL (Dube et al., 1986; Raimondi et al., 1988; Kagan et al., 1989; Uckun et al., 1989; Parket et al., 1992; Secker-Walker et al., 1992; Martin et al., 1996; Rack et al., 1997; Forestier et al., 1998; Heerema et al., 1998; Lai et al., 2000; Schneider et al., 2000; Kahl et al., 2001; Pedersen et al., 2001; Thomas et al., 2001; Mancini et al., 2002; Nordgren et al., 2002; Kristensen et al., 2003; Barber et al., 2004; Speleman et al., 2005; Stergianou et al., 2005; Cauwelier et al., 2006; Reichard et al., 2006; van Grotel et al., 2006; Strefford et al., 2007; Kwon et al., 2009; Le Noir et al., 2012; Setoodeh & Zhang, 2012; Grossmann et al., 2013; Park et al., 2014; Safavi et al., 2015; Gindina T. three personal unpublished cases).

A t(7;14)(q34;q24) TLX1/TRB was found in 21 cases of T-ALL (Kahl et al., 2001; Cauwelier et al., 2006; Le Noir et al., 2012; Raimondi et al., 1988).

In exceptional cases, a t(10;14) or a t(7;10) was found in T-prolymphocytic leukemia and ataxia telangiectasia (Rack et al., 1997), and, more surprisingly, in chronic lymphocytic leukemia, a B-cell disease (Delhomme-Bachy et al., 1992; Lu et al., 2006).

### Phenotype/cell stem origin

T lineage. The gene expression pattern of TLX1-expressing lymphoblasts is similar to that of early cortical thymocytes, compatible with a leukemic arrest at the stage of the early cortical thymocyte (virtually all arrested at the early cortical (CD1+) CD4+ CD8+ "double-positive" stage of thymocyte development).

### Epidemiology

Found in 5% of pediatric T-ALL (0.3% of all pediatric ALL) and 30% of adult T-ALL cases. Median age was 24 years (range 4-58). Sex ratio was 3 male : 1 female patients.

### Clinics

Organomegaly with marked hepatosplenomegaly, lymphadenopathy, mediastinal mass, high WBC count (100 to 200 X 10<sup>9</sup>/l) sometimes with anemia.

### Cytology

High leukocyte count, very high circulating and central blast cell count.

### Prognosis

TLX1 expression has been linked with a favorable prognosis and low risk of relapse in children and adults (van Vlierberghe et al., 2012). TLX1+ patients have a 92% probability of survival at 5 years. The lack of expression of anti-apoptotic genes in this stage of thymocyte development (and in TLX1-expressing lymphoblasts) leads to a high responsiveness to drug-induced apoptosis.

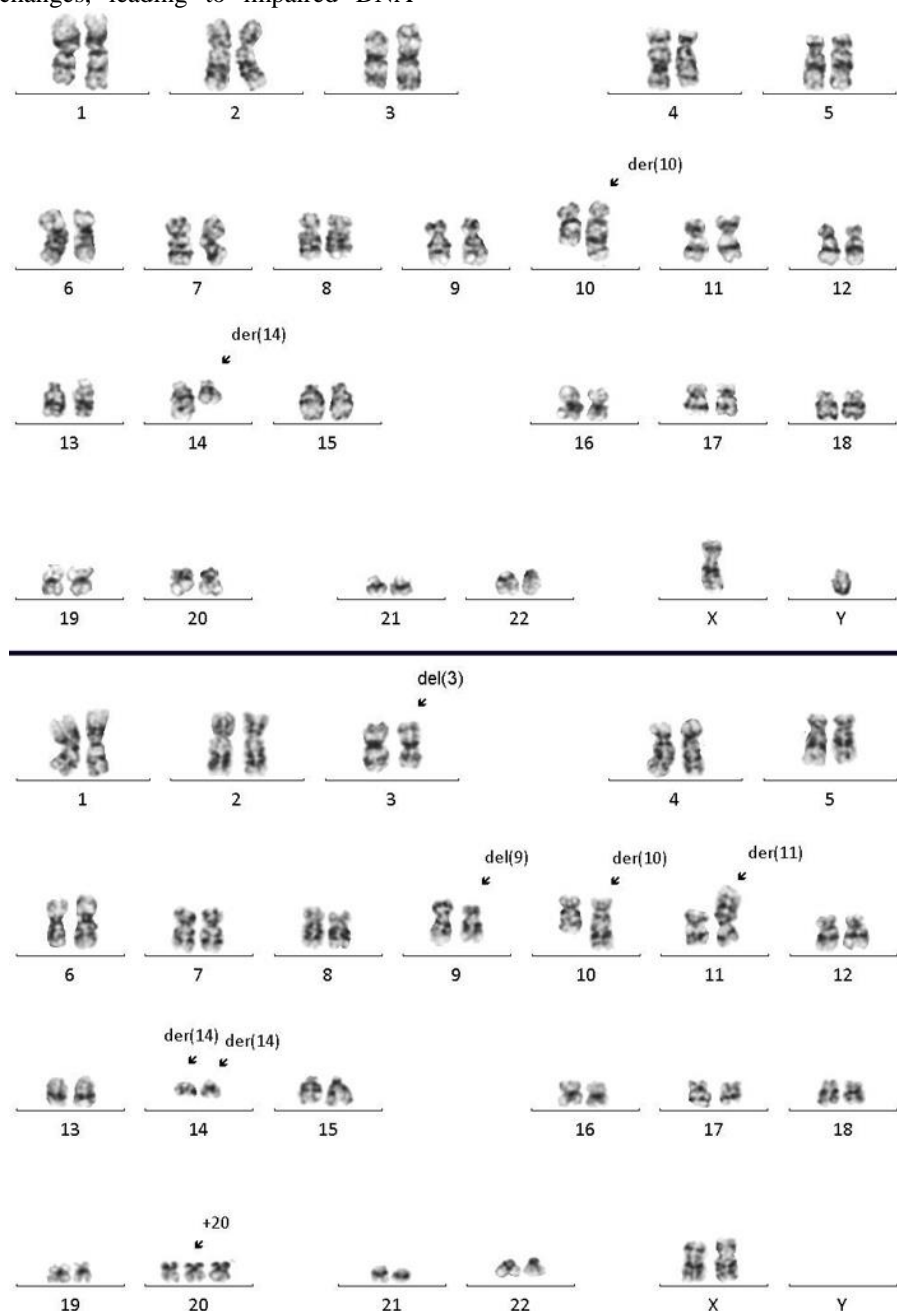
## Genetics

### Note

Both translocations t(10;14)(q24;q11.2) and t(7;10)(q34;q24) are insufficient to initiate malignancy in mice: activation of other mutant genes, including NOTCH1, is found in most TLX1+ T-ALL. This finding suggests that multiple cooperating changes, leading to impaired DNA

repair, lead to T-cell differentiation arrest and leukemogenesis.

## Cytogenetics



Top: Translocation t(10;14)(q24;q11) in a 32-year-old male with T-ALL and OS 33 months+ - Courtesy Tatiana Gindina. Bottom: Complex karyotype with two translocations t(10;14)(q24;q11) and t(11;14)(p13;q11), deletions of 3q, 9p, trisomy 20 in a 31-year-old female with T-ALL and OS 37 months - Courtesy Tatiana Gindina.

### Additional anomalies

Additional chromosome anomalies were observed in about half of the cases (Dube et al., 1986; Raimondi et al., 1988; Kagan et al., 1989; Rack et al., 1997; Forestier et al., 1998; Lai et al., 2000; Schneider et al., 2000; Kahl et al., 2001; Pedersen et al., 2001;

Mancini et al., 2002; Nordgren et al., 2002; Kristensen et al., 2003; Barber et al., 2004; Speleman et al., 2005; Stergianou et al., 2005; Cauwelier et al., 2006; Reichard et al., 2006; Strefford et al., 2007; Le Noir et al., 2012; Setoodeh & Zhang, 2012; Grossmann et al., 2013; Safavi et al., 2015; Gindina T., three personal cases). The most common of them

t(10;14)(q24;q11) or t(7;10) TLX1/TRD or TRB

are deletions of 9p (17%), 6q (13%), and 12p (9%). Deletions of 11p, 6p, 3q, 7q, 13q were less common. Trisomy 8 was present in 6 (7%) patients, and trisomy 20 was in 4 (4,5%) patients. Trisomies of other chromosomes were detected very seldom. In 34 (38%) cases the translocations t(10;14) and t(7;10) were a part of a complex karyotype.

## Genes involved and proteins

### ***TLX1 (T-cell leukemia homeobox 1)***

#### **Location**

10q24.31

#### **Note**

Alias HOX11 (homeobox 11) alias TCL3 (T-cell leukemia 3).

#### **DNA/RNA**

spans over 7 kb, 3 exons, mRNA 7 kb

#### **Protein**

The homeobox gene TLX1 (HOX11) encodes for TLX1, a homeodomain-containing transcription factor, nuclear transcription factor that belongs to the NK-linked or NK-like subfamily of homeobox genes. TLX1 is the founding member of a family of HOX genes, homeoprotein, 61 amino acids, nuclear localization. It binds to the DNA sequence 5'-GGCGGTAAGTGG-3'. The encoded protein is required and critical for normal development of the spleen during embryogenesis and is also involved in specification of neuronal cell fate. TLX1 is not normally expressed in adult tissues at levels detectable by routine Northern analysis.

TLX-1 leukemias show specific cooperating mutations rarely present in non-TLX-induced leukemias, including the NUP214 / ABL1 fusion oncogene and mutations in the PTPN2, Wilms tumor 1 (WT1), and PHF6 tumor suppressors (van Vlierberghe et al., 2012).

### ***TRD (T cell Receptor Delta)***

#### **Location**

14q11.2

#### **Note**

or TRB in the case of a 7q34-36 involvement

#### **Protein**

T-cell receptor

## Result of the chromosomal anomaly

### ***Fusion protein***

#### **Description**

Both translocations place TLX1 under the control of strong enhancers in the T-cell receptor loci. The t(10;14) translocation places the TLX1 coding region under the transcriptional control of the TCR

delta receptor (TRD)( 14q11.2) promoter, the t(7;10) translocation places the TLX1 coding region under the transcriptional control of the TCR beta locus (TRB)(7q34-36), both leading to increased expression of TLX1 in T-cells. The t(10;14) can be detected by PCR, and a dual-color FISH probe is often used to detect HOX11 translocations on 10q24. However, TLX1 overexpression in leukemic blasts has been observed in the absence of 10q24 rearrangement in as many as 50% of T-ALL cases.

#### **Oncogenesis**

These genetic aberrations induce aberrant and abundant TLX1 expression in T- lineage cells bearing the translocation. In addition, it was recently proposed that the unique cortical thymic maturation arrest in TLX-induced leukemias may be related to the binding of TLX1- ETS1 complexes to TCRA enhancer sequences, with the consequent down regulation of TCRA gene rearrangement and expression (van Vlierberghe et al., 2012).

TLX1 overexpressing lymphoblasts are arrested at the stage of beta-selection in the thymocyte development. Leukemogenesis results from decreased cell death and increased proliferation of immature TLX1 expressing thymocytes, in the absence of normal DNA repair systems. The lack of anti-apoptotic actors in this stage explains the high responsiveness to chemotherapy, and the associated excellent outcome.

## References

- . Birth Defects Cytogenet Cell Genet. 1974;13(3):1-216
- 1000 Genomes Project Consortium, Auton A, Brooks LD, Durbin RM, Garrison EP, Kang HM, Korbel JO, Marchini JL, McCarthy S, McVean GA, Abecasis GR. A global reference for human genetic variation Nature 2015 Oct 1;526(7571):68-74
- A PROPOSED standard system of nomenclature of human mitotic chromosomes. Lancet 1960 May 14;1(7133):1063-5 PubMed PMID: 13857542
- Amberger JS, Bocchini CA, Schiettecatte F, Scott AF, Hamosh A. OMIM.org: Online Mendelian Inheritance in Man (OMIM), an online catalog of human genes and genetic disorders Nucleic Acids Res
- Aurias A, Rimbaut C, Buffe D, Dubousset J, Mazabraud A. [Translocation of chromosome 22 in Ewing's sarcoma] C R Seances Acad Sci III 1983;296(23):1105-7
- BEADLE GW. Genetics and metabolism in Neurospora Physiol Rev 1945 Oct;25:643-63
- Babiceanu M, Qin F, Xie Z, Jia Y, Lopez K, Janus N, Facemire L, Kumar S, Pang Y, Qi Y, Lazar IM, Li H. Recurrent chimeric fusion RNAs in non-cancer tissues and cells Nucleic Acids Res 2016 Apr 7;44(6):2859-72
- Baccelli I, Schneeweiss A, Riethdorf S, Stenzinger A, Schillert A, Vogel V, Klein C, Saini M, Bäuerle T, Wallwiener M, Holland-Letz T, Höfner T, Sprick M, Scharpf M, Marmé F, Sinn HP, Pantel K, Weichert W, Trumpp A. Identification of a population of blood circulating tumor cells from breast cancer patients that initiates metastasis in a xenograft assay Nat Biotechnol 2013 Jun;31(6):539-44

- Barr FG, Nauta LE, Davis RJ, Schäfer BW, Nycum LM, Biegel JA. In vivo amplification of the PAX3-FKHR and PAX7-FKHR fusion genes in alveolar rhabdomyosarcoma *Hum Mol Genet* 1996 Jan;5(1):15-21
- Benson DA, Clark K, Karsch-Mizrachi I, Lipman DJ, Ostell J, Sayers EW. GenBank Nucleic Acids Res 2015 Jan;43(Database issue):D30-5
- Benton D. Recent changes in the GenBank On-line Service *Nucleic Acids Res* 1990 Mar 25;18(6):1517-20
- Berger R, Bernheim A, Weh HJ, Flandrin G, Daniel MT, Brouet JC, Colbert N. A new translocation in Burkitt's tumor cells *Hum Genet* 1979;53(1):111-2
- Bernard O, Lecointe N, Jonveaux P, Souyri M, Mauchauffé M, Berger R, Larsen CJ, Mathieu-Mahul D. Two site-specific deletions and t(1;14) translocation restricted to human T-cell acute leukemias disrupt the 5' part of the tal-1 gene *Oncogene* 1991 Aug;6(8):1477-88
- Bernheim A, Huret JL, Guillaud-Bataille M, Brison O, Couturiers J; Groupe Français de Cytogéné Oncologique. [Cytogenetics, cytogenomics and cancer: 2004 update] *Bull Cancer* 2004 Jan;91(1):29-43
- Beroukhim R, Mermel CH, Porter D, Wei G, Raychaudhuri S, Donovan J, Barretina J, Boehm JS, Dobson J, Urashima M, Mc Henry KT, Pinchback RM, Ligon AH, Cho YJ, Haery L, Greulich H, Reich M, Winckler W, Lawrence MS, Weir BA, Tanaka KE, Chiang DY, Bass AJ, Loo A, Hoffman C, Prensner J, Liefeld T, Gao Q, Yecies D, Signoretti S, Maher E, Kaye FJ, Sasaki H, Tepper JE, Fletcher JA, Taberero J, Baselga J, Tsao MS, Demicheli F, Rubin MA, Janne PA, Daly MJ, Nucera C, Levine RL, Ebert BL, Gabriel S, Rustgi AK, Antonescu CR, Ladanyi M, Letai A, Garraway LA, Loda M, Beer DG, True LD, Okamoto A, Pomeroy SL, Singer S, Golub TR, Lander ES, Getz G, Sellers WR, Meyerson M. The landscape of somatic copy-number alteration across human cancers *Nature* 2010 Feb 18;463(7283):899-905
- Boveri T... Zur Frage der Entstehung maligner Tumoren 1914 Gustav Fischer
- Brookes AJ, Robinson PN. Human genotype-phenotype databases: aims, challenges and opportunities *Nat Rev Genet* 2015 Dec;16(12):702-15
- Burks C, Cassidy M, Cinkosky MJ, Cumella KE, Gilna P, Hayden JE, Keen GM, Kelley TA, Kelly M, Kristofferson D, et al. GenBank Nucleic Acids Res 1991 Apr 25;19 Suppl:2221-5
- Burks C, Fickett JW, Goad WB, Kanehisa M, Lewitter FI, Rindone WP, Swindell CD, Tung CS, Bilofsky HS. The GenBank nucleic acid sequence database *Comput Appl Biosci* 1985 Dec;1(4):225-33
- Cai H, Gupta S, Rath P, Ai N, Baudis M. arrayMap 2014: an updated cancer genome resource *Nucleic Acids Res* 2015 Jan;43(Database issue):D825-30
- Campbell PJ, Stephens PJ, Pleasance ED, O'Meara S, Li H, Santarius T, Stebbings LA, Leroy C, Edkins S, Hardy C, Teague JW, Menzies A, Goodhead I, Turner DJ, Clee CM, Quail MA, Cox A, Brown C, Durbin R, Hurles ME, Edwards PA, Bignell GR, Stratton MR, Futreal PA. Identification of somatically acquired rearrangements in cancer using genome-wide massively parallel paired-end sequencing *Nat Genet* 2008 Jun;40(6):722-9
- Cancer Genome Atlas Research Network. Integrated genomic analyses of ovarian carcinoma *Nature* 2011 Jun 29;474(7353):609-15
- Cao Q, Zhou M, Wang X, Meyer CA, Zhang Y, Chen Z, Li C, Liu XS. CaSNP: a database for interrogating copy number alterations of cancer genome from SNP array data *Nucleic Acids Res* 2011 Jan;39(Database issue):D968-74
- Caspersson T, Zech L, Modest EJ. Fluorescent labeling of chromosomal DNA: superiority of quinacrine mustard to quinacrine Science 1970 Nov 13;170(3959):762
- Chen CW, Koche RP, Sinha AU, Deshpande AJ, Zhu N, Eng R, Doench JG, Xu H, Chu SH, Qi J, Wang X, Delaney C, Bernt KM, Root DE, Hahn WC, Bradner JE, Armstrong SA. DOT1L inhibits SIRT1-mediated epigenetic silencing to maintain leukemic gene expression in MLL-rearranged leukemia *Nat Med* 2015 Apr;21(4):335-43
- Chin L, Hahn WC, Getz G, Meyerson M.. Making sense of cancer genomic data. *Genes Dev*. 2011 Mar 15;25(6):534-55. doi: 10.1101/gad.2017311.
- Clough E, Barrett T. The Gene Expression Omnibus Database Methods Mol Biol 2016;1418:93-110
- Commo F, Ferté C, Soria JC, Friend SH, André F, Guinney J. Impact of centralization on aCGH-based genomic profiles for precision medicine in oncology *Ann Oncol* 2015 Mar;26(3):582-8
- Cook CE, Bergman MT, Finn RD, Cochrane G, Birney E, Apweiler R. The European Bioinformatics Institute in 2016: Data growth and integration *Nucleic Acids Res* 2016 Jan 4;44(D1):D20-6
- Cooper DN, Krawczak M. Human Gene Mutation Database *Hum Genet* 1996 Nov;98(5):629
- Crowley E, Di Nicolantonio F, Loupakis F, Bardelli A. Liquid biopsy: monitoring cancer-genetics in the blood *Nat Rev Clin Oncol* 2013 Aug;10(8):472-84
- Dawson MA, Prinjha RK, Dittmann A, Giotopoulos G, Bantscheff M, Chan WJ, Robson SC, Chung CW, Hopf C, Savitski MM, Huthmacher C, Gudgin E, Lugo D, Beinke S, Chapman TD, Roberts EJ, Soden PE, Auger KR, Mirquet O, Doehner K, Delwel R, Burnett AK, Jeffrey P, Drewes G, Lee K, Huntly BJ, Kouzarides T. Inhibition of BET recruitment to chromatin as an effective treatment for MLL-fusion leukaemia *Nature* 2011 Oct 2;478(7370):529-33
- De Braekeleer E, Douet-Guilbert N, De Braekeleer M. Genetic diagnosis in malignant hemopathies: from cytogenetics to next-generation sequencing *Expert Rev Mol Diagn* 2014 Mar;14(2):127-9
- Deng M, Brägelmann J, Schultze JL, Perner S. Web-TCGA: an online platform for integrated analysis of molecular cancer data sets *BMC Bioinformatics* 2016 Feb 6;17:72
- Diehl AG, Boyle AP. Deciphering ENCODE *Trends Genet* 2016 Apr;32(4):238-49
- Dorkeld F, Bernheim A, Dessen P, Huret JL. A database on cytogenetics in haematology and oncology *Nucleic Acids Res* 1999 Jan 1;27(1):353-4
- Druker BJ, Sawyers CL, Kantarjian H, Resta DJ, Reese SF, Ford JM, Capdeville R, Talpaz M. Activity of a specific inhibitor of the BCR-ABL tyrosine kinase in the blast crisis of chronic myeloid leukemia and acute lymphoblastic leukemia with the Philadelphia chromosome *N Engl J Med* 2001 Apr 5;344(14):1038-42
- Druker BJ, Talpaz M, Resta DJ, Peng B, Buchdunger E, Ford JM, Lydon NB, Kantarjian H, Capdeville R, Ohno-Jones S, Sawyers CL. Efficacy and safety of a specific inhibitor of the BCR-ABL tyrosine kinase in chronic myeloid leukemia *N Engl J Med* 2001 Apr 5;344(14):1031-7
- Fillmore CM, Xu C, Desai PT, Berry JM, Rowbotham SP, Lin YJ, Zhang H, Marquez VE, Hammerman PS, Wong KK, Kim CF. EZH2 inhibition sensitizes BRG1 and EGFR mutant lung tumours to TopoII inhibitors *Nature* 2015 Apr 9;520(7546):239-42
- Finn RD, Coghill P, Eberhardt RY, Eddy SR, Mistry J, Mitchell AL, Potter SC, Punta M, Qureshi M, Sangrador-

- Vegas A, Salazar GA, Tate J, Bateman A. The Pfam protein families database: towards a more sustainable future *Nucleic Acids Res* 2016 Jan 4;44(D1):D279-85
- Firth HV, Richards SM, Bevan AP, Clayton S, Corpes M, Rajan D, Van Vooren S, Moreau Y, Pettett RM, Carter NP. DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources *Am J Hum Genet* 2009 Apr;84(4):524-33
- Fishilevich S, Zimmerman S, Kohn A, Iny Stein T, Olender T, Kolker E, Safran M, Lancet D. Genic insights from integrated human proteomics in GeneCards Database (Oxford) 2016 Apr 5;2016
- Fletcher CD. The evolving classification of soft tissue tumours - an update based on the new 2013 WHO classification *Histopathology* 2014 Jan;64(1):2-11
- Fokkema IF, Taschner PE, Schaafsma GC, Celli J, Laros JF, den Dunnen JT. LOVD v. 2.0: the next generation in gene variant databases. *Hum Mutat*. May;32(5):557-63
- Forbes SA, Beare D, Gunasekaran P, Leung K, Bindal N, Boutselakis H, Ding M, Bamford S, Cole C, Ward S, Kok CY, Jia M, De T, Teague JW, Stratton MR, McDermott U, Campbell PJ. COSMIC: exploring the world's knowledge of somatic mutations in human cancer *Nucleic Acids Res* 2015 Jan;43(Database issue):D805-11
- Frenkel-Morgenstern M, Gorohovski A, Vucenovic D, Maestre L, Valencia A. ChiTaRS 2.1--an improved database of the chimeric transcripts and RNA-seq data with novel sense-antisense chimeric RNA transcripts *Nucleic Acids Res*
- Fukuhara S, Rowley JD, Variakojis D, Golomb HM. Chromosome abnormalities in poorly differentiated lymphocytic lymphoma *Cancer Res* 1979 Aug;39(8):3119-28
- Futreal PA, Coin L, Marshall M, Down T, Hubbard T, Wooster R, Rahman N, Stratton MR. A census of human cancer genes *Nat Rev Cancer* 2004 Mar;4(3):177-83
- Gaudet P, Michel PA, Zahn-Zabal M, Cusin I, Duek PD, Evalet O, Gateau A, Gleizes A, Pereira M, Teixeira D, Zhang Y, Lane L, Bairoch A. The neXtProt knowledgebase on human proteins: current status *Nucleic Acids Res* 2015 Jan;43(Database issue):D764-70
- Gelsi-Boyer V, Trouplin V, Adélaïde J, Aceto N, Remy V, Pinson S, Houdayer C, Arnoulet C, Sainty D, Bentires-Alj M, Olschwang S, Vey N, Mozziconacci MJ, Birnbaum D, Chaffanet M. Genome profiling of chronic myelomonocytic leukemia: frequent alterations of RAS and RUNX1 genes *BMC Cancer* 2008 Oct 16;8:299
- Gingeras TR. Implications of chimaeric non-co-linear transcripts *Nature* 2009 Sep 10;461(7261):206-11
- Goldman M, Craft B, Swatloski T, Cline M, Morozova O, Diekhans M, Haussler D, Zhu J. The UCSC Cancer Genomics Browser: update 2015 *Nucleic Acids Res* 2015 Jan;43(Database issue):D812-7
- Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Genenames.org: the HGNC resources in 2015 *Nucleic Acids Res*
- Gundem G, Perez-Llamas C, Jene-Sanz A, Kedzierska A, Islam A, Deu-Pons J, Furney SJ, Lopez-Bigas N. IntOGen: integration and data mining of multidimensional oncogenomic data *Nat Methods* 2010 Feb;7(2):92-3
- Harrison SM, Riggs ER, Maglott DR, Lee JM, Azzariti DR, Niehaus A, Ramos EM, Martin CL, Landrum MJ, Reh HL. Using ClinVar as a Resource to Support Variant Interpretation *Curr Protoc Hum Genet* 2016 Apr 1;89:8
- Hedegaard J, Thorsen K, Lund MK, Hein AM, Hamilton-Dutoit SJ, Vang S, Nordentoft I, Birkenkamp-Demtröder K, Kruhøffer M, Hager H, Knudsen B, Andersen CL, Sørensen KD, Pedersen JS, Ørntoft TF, Dyrskjødt L. Next-generation sequencing of RNA and DNA isolated from paired fresh-frozen and formalin-fixed paraffin-embedded samples of human cancer and normal tissue *PLoS One* 2014 May 30;9(5):e98187
- Heim S, Mandahl N, Kristoffersson U, Mitelman F, Rser B, Rydholm A, Willén H. Reciprocal translocation t(3;12)(q27;q13) in lipoma *Cancer Genet Cytogenet* 1986 Dec;23(4):301-4
- Hnisz D, Weintraub AS, Day DS, Valton AL, Bak RO, Li CH, Goldmann J, Lajoie BR, Fan ZP, Sigova AA, Reddy J, Borges-Rivera D, Lee TI, Jaenisch R, Porteus MH, Dekker J, Young RA. Activation of proto-oncogenes by disruption of chromosome neighborhoods *Science* 2016 Mar 25;351(6280):1454-8
- Hokland P, Ommen HB. Towards individualized follow-up in adult acute myeloid leukemia in remission *Blood* 2011 Mar 3;117(9):2577-84
- Hornbeck PV, Zhang B, Murray B, Kornhauser JM, Latham V, Skrzypek E. PhosphoSitePlus, 2014: mutations, PTMs and recalibrations *Nucleic Acids Res* 2015 Jan;43(Database issue):D512-20
- Huret JL, Ahmad M, Arsaban M, Bernheim A, Cigna J, Desangles F, Guignard JC, Jacquemot-Perbal MC, Labarussias M, Leberre V, Malo A, Morel-Pair C, Mossafa H, Potier JC, Texier G, Viguié F, Yau Chun Wan-Senon S, Zasadzinski A, Dessen P. Atlas of genetics and cytogenetics in oncology and haematology in 2013 *Nucleic Acids Res* 2013 Jan;41(Database issue):D920-4
- Iafrate AJ, Feuk L, Rivera MN, Listewnik ML, Donahoe PK, Qi Y, Scherer SW, Lee C. Detection of large-scale variation in the human genome *Nat Genet* 2004 Sep;36(9):949-51
- International HapMap 3 Consortium, Altshuler DM, Gibbs RA, Peltonen L, Altshuler DM, Gibbs RA, Peltonen L, Dermitzakis E, Schaffner SF, Yu F, Peltonen L, Dermitzakis E, Bonnen PE, Altshuler DM, Gibbs RA, de Bakker PI, Deloukas P, Gabriel SB, Gwilliam R, Hunt S, Inouye M, Jia X, Palotie A, Parkin M, Whittaker P, Yu F, Chang K, Hawes A, Lewis LR, Ren Y, Wheeler D, Gibbs RA, Muzny DM, Barnes C, Darvishi K, Hurler M, Korn JM, Kristiansson K, Lee C, McCarroll SA, Nemesh J, Dermitzakis E, Keinan A, Montgomery SB, Pollack S, Price AL, Soranzo N, Bonnen PE, Gibbs RA, Gonzaga-Jauregui C, Keinan A, Price AL, Yu F, Anttila V, Brodeur W, Daly MJ, Leslie S, McVean G, Moutsianas L, Nguyen H, Schaffner SF, Zhang Q, Ghorri MJ, McGinnis R, McLaren W, Pollack S, Price AL, Schaffner SF, Takeuchi F, Grossman SR, Shlyakhter I, Hostetter EB, Sabeti PC, Adebamowo CA, Foster MW, Gordon DR, Licinio J, Manca MC, Marshall PA, Matsuda I, Ngare D, Wang VO, Reddy D, Rotimi CN, Royal CD, Sharp RR, Zeng C, Brooks LD, McEwen JE. Integrating common and rare genetic variation in diverse human populations *Nature* 2010 Sep 2;467(7311):52-8
- Joensuu H. Adjuvant treatment of GIST: patient selection and treatment strategies *Nat Rev Clin Oncol* 2012 Apr 24;9(6):351-8
- Kallioniemi A, Kallioniemi OP, Sudar D, Rutovitz D, Gray JW, Waldman F, Pinkel D. Comparative genomic hybridization for molecular cytogenetic analysis of solid tumors *Science* 1992 Oct 30;258(5083):818-21
- Kalyana-Sundaram S, Shankar S, Deroo S, Iyer MK, Palanisamy N, Chinnaiyan AM, Kumar-Sinha C. Gene fusions associated with recurrent amplicons represent a class of passenger aberrations in breast cancer Neoplasia 2012 Aug;14(8):702-8



- Karabacak NM, Spuhler PS, Fachin F, Lim EJ, Pai V, Ozkumur E, Martel JM, Kojic N, Smith K, Chen PI, Yang J, Hwang H, Morgan B, Trautwein J, Barber TA, Stott SL, Maheswaran S, Kapur R, Haber DA, Toner M. Microfluidic, marker-free isolation of circulating tumor cells from blood samples *Nat Protoc* 2014 Mar;9(3):694-710
- Kim N, Kim P, Nam S, Shin S, Lee S. ChimerDB--a knowledgebase for fusion sequences *Nucleic Acids Res* 2006 Jan 1;34(Database issue):D21-4
- Kim P, Yoon S, Kim N, Lee S, Ko M, Lee H, Kang H, Kim J, Lee S. ChimerDB 2.0--a knowledgebase for fusion genes updated *Nucleic Acids Res*
- Kim TM, Xi R, Luquette LJ, Park RW, Johnson MD, Park PJ. Functional genomic analysis of chromosomal aberrations in a compendium of 8000 cancer genomes *Genome Res* 2013 Feb;23(2):217-27
- Klijn C, Durinck S, Stawiski EW, Haverty PM, Jiang Z, Liu H, Degenhardt J, Mayba O, Gnad F, Liu J, Pau G, Reeder J, Cao Y, Mukhyala K, Selvaraj SK, Yu M, Zynda GJ, Brauer MJ, Wu TD, Gentleman RC, Manning G, Yauch RL, Bourgon R, Stokoe D, Modrusan Z, Neve RM, de Sauvage FJ, Settleman J, Seshagiri S, Zhang Z. A comprehensive transcriptional portrait of human cancer cell lines *Nat Biotechnol* 2015 Mar;33(3):306-12
- Klonowska K, Czubak K, Wojciechowska M, Handschuh L, Zmienko A, Figlerowicz M, Dams-Kozłowska H, Kozłowski P. Oncogenomic portals for the visualization and analysis of genome-wide cancer data *Oncotarget* 2016 Jan 5;7(1):176-92
- Kohno T, Tsuta K, Tsuchihara K, Nakaoku T, Yoh K, Goto K. RET fusion gene: translation to personalized lung cancer therapy *Cancer Sci* 2013 Nov;104(11):1396-400
- Løv M, Thomassen GO, Bakken AC, Celestino R, Fioretos T, Lind GE, Lothe RA, Skotheim RI. Fusion gene microarray reveals cancer type-specificity among fusion genes *Genes Chromosomes Cancer* 2011 May;50(5):348-57
- Lawler M, Siu LL, Rehm HL, Chanock SJ, Alterovitz G, Burn J, Calvo F, Lacombe D, Teh BT, North KN, Sawyers CL; Clinical Working Group of the Global Alliance for Genomics and Health (GA4GH). All the World's a Stage: Facilitating Discovery Science and Improved Cancer Care through the Global Alliance for Genomics and Health *Cancer Discov* 2015 Nov;5(11):1133-6
- Leary RJ, Kinde I, Diehl F, Schmidt K, Clouser C, Duncan C, Antipova A, Lee C, McKernan K, De La Vega FM, Kinzler KW, Vogelstein B, Diaz LA Jr, Velculescu VE. Development of personalized tumor biomarkers using massively parallel sequencing *Sci Transl Med* 2010 Feb 24;2(20):20ra14
- Lee HJ, Thompson JE, Wang ES, Wetzler M. Philadelphia chromosome-positive acute lymphoblastic leukemia: current treatment and future perspectives *Cancer* 2011 Apr 15;117(8):1583-94
- Möller E, Hornick JL, Magnusson L, Veerla S, Domanski HA, Mertens F. FUS-CREB3L2/L1-positive sarcomas show a specific gene expression profile with upregulation of CD24 and FOXL1 *Clin Cancer Res* 2011 May 1;17(9):2646-56
- MacDonald JR, Ziman R, Yuen RK, Feuk L, Scherer SW. The Database of Genomic Variants: a curated collection of structural variation in the human genome *Nucleic Acids Res* 2014 Jan;42(Database issue):D986-92
- Maher CA, Kumar-Sinha C, Cao X, Kalyana-Sundaram S, Han B, Jing X, Sam L, Barrette T, Palanisamy N, Chinnaiyan AM. Transcriptome sequencing to detect gene fusions in cancer *Nature* 2009 Mar 5;458(7234):97-101
- Maher CA, Palanisamy N, Brenner JC, Cao X, Kalyana-Sundaram S, Luo S, Khrebtkova I, Barrette TR, Grasso C, Yu J, Lonigro RJ, Schroth G, Kumar-Sinha C, Chinnaiyan AM. Chimeric transcript discovery by paired-end transcriptome sequencing *Proc Natl Acad Sci U S A* 2009 Jul 28;106(30):12353-8
- Malik R, Khan AP, Asangani IA, Cielik M, Prensner JR, Wang X, Iyer MK, Jiang X, Borkin D, Escara-Wilke J, Stender R, Wu YM, Niknafs YS, Jing X, Qiao Y, Palanisamy N, Kunju LP, Krishnamurthy PM, Yocum AK, Mellacheruvu D, Nesvizhskii AI, Cao X, Dhanasekaran SM, Feng FY, Grembecka J, Cierpicki T, Chinnaiyan AM. Targeting the MLL complex in castration-resistant prostate cancer *Nat Med* 2015 Apr;21(4):344-52
- Mark J, Dahlenfors R, Ekedahl C, Stenman G. The mixed salivary gland tumor A normally benign human neoplasm frequently showing specific chromosomal abnormalities. *Cancer Genetics and Cytogenetics* 1980 2, 231-24
- Martincorena I, Campbell PJ. Somatic mutation in cancer and normal cells *Science* 2015 Sep 25;349(6255):1483-9
- McCabe MT, Ott HM, Ganji G, Korenchuk S, Thompson C, Van Aller GS, Liu Y, Graves AP, Della Pietra A 3rd, Diaz E, LaFrance LV, Mellinger M, Duquenne C, Tian X, Kruger RG, McHugh CF, Brandt M, Miller WH, Dhanak D, Verma SK, Tummino PJ, Creasy CL. EZH2 inhibition as a therapeutic strategy for lymphoma with EZH2-activating mutations *Nature* 2012 Dec 6;492(7427):108-12
- Mertens F, Johansson B, Fioretos T, Mitelman F. The emerging complexity of gene fusions in cancer *Nat Rev Cancer* 2015 Jun;15(6):371-81
- Meyer C, Hofmann J, Burmeister T, Gröger D, Park TS, Emerenciano M, Pombo de Oliveira M, Renneville A, Villares P, Macintyre E, Cavé H, Clappier E, Mass-Malo K, Zuna J, Trka J, De Braekeleer E, De Braekeleer M, Oh SH, Tsaur G, Fechina L, van der Velden VH, van Dongen JJ, Delabesse E, Binato R, Silva ML, Kustanovich A, Aleinikova O, Harris MH, Lund-Aho T, Juvonen V, Heidenreich O, Vormoor J, Choi WW, Jarosova M, Kolenova A, Bueno C, Menendez P, Wehner S, Eckert C, Talmant P, Tondeur S, Lippert E, Launay E, Henry C, Ballerini P, Lapillone H, Callanan MB, Cayuela JM, Herbaux C, Cazzaniga G, Kakadiya PM, Bohlander S, Ahlmann M, Choi JR, Gameiro P, Lee DS, Krauter J, Cornillet-Lefebvre P, Te Kronnie G, Schäfer BW, Kubetzko S, Alonso CN, zur Stadt U, Sutton R, Venn NC, Izraeli S, Trakhtenbrot L, Madsen HO, Archer P, Hancock J, Cerveira N, Teixeira MR, Lo Nigro L, Möricke A, Stanulla M, Schrappe M, Sedék L, Szczepański T, Zwaan CM, Coenen EA, van den Heuvel-Eibrink MM, Strehl S, Dworzak M, Panzer-Grümayer R, Dingermann T, Klingebiel T, Marschalek R. The MLL recombinome of acute leukemias in 2013 *Leukemia* 2013 Nov;27(11):2165-76
- Meyer C, Kowarz E, Hofmann J, Renneville A, Zuna J, Trka J, Ben Abdelali R, Macintyre E, De Braekeleer E, De Braekeleer M, Delabesse E, de Oliveira MP, Cavé H, Clappier E, van Dongen JJ, Balgobind BV, van den Heuvel-Eibrink MM, Beverloo HB, Panzer-Grümayer R, Teigler-Schlegel A, Harbott J, Kjeldsen E, Schnittger S, Koehl U, Gruhn B, Heidenreich O, Chan LC, Yip SF, Krzywinski M, Eckert C, Möricke A, Schrappe M, Alonso CN, Schäfer BW, Krauter J, Lee DA, Zur Stadt U, Te Kronnie G, Sutton R, Izraeli S, Trakhtenbrot L, Lo Nigro L, Tsaur G, Fechina L, Szczepański T, Strehl S, Ilencikova D, Molkenin M, Burmeister T, Dingermann T, Klingebiel T, Marschalek R. New insights to the MLL recombinome of acute leukemias *Leukemia* 2009 Aug;23(8):1490-9
- Minikel EV, Vallabh SM, Lek M, Estrada K, Samocha KE, Sathirapongsasuti JF, McLean CY, Tung JY, Yu LP, Gambetti P, Blevins J, Zhang S, Cohen Y, Chen W, Yamada M, Hamaguchi T, Sanjo N, Mizusawa H, Nakamura Y, Kitamoto T, Collins SJ, Boyd A, Will RG, Knight R, Ponto C, Zerr I, Kraus TF, Eigenbrod S, Giese A, Calero M, de Pedro-Cuesta J, Haik S, Laplanche JL, Bouaziz-Amar E, Brandel

- JP, Capellari S, Parchi P, Poleggi A, Ladogana A, O'Donnell-Luria AH, Karczewski KJ, Marshall JL, Boehnke M, Laakso M, Mohlke KL, Kähler A, Chambert K, McCarroll S, Sullivan PF, Hultman CM, Purcell SM, Sklar P, van der Lee SJ, Rozemuller A, Jansen C, Hofman A, Kraaij R, van Rooij JG, Ikram MA, Uitterlinden AG, van Duijn CM; Exome Aggregation Consortium (ExAC), Daly MJ, MacArthur DG. Quantifying prion disease penetrance using large population control cohorts *Sci Transl Med* 2016 Jan 20;8(322):322ra9
- Mitchell A, Chang HY, Daugherty L, Fraser M, Hunter S, Lopez R, McAnulla C, McMenamin C, Nuka G, Pesseat S, Sangrador-Vegas A, Scheremetjew M, Rato C, Yong SY, Bateman A, Punta M, Attwood TK, Sigrist CJ, Redaschi N, Rivoire C, Xenarios I, Kahn D, Guyot D, Bork P, Letunic I, Gough J, Oates M, Haft D, Huang H, Natale DA, Wu CH, Orengo C, Sillitoe I, Mi H, Thomas PD, Finn RD. The InterPro protein families database: the classification resource after 15 years *Nucleic Acids Res* 2015 Jan;43(Database issue):D213-21
- Mitelman F, Johansson B, Mertens F. Mitelman database of chromosome aberrations and genes fusions in Cancer
- Miyoshi I, Hiraki S, Kimura I, Miyamoto K, Sato J. 2/8 translocation in a Japanese Burkitt's lymphoma *Experientia* 1979 Jun 15;35(6):742-3
- Mullighan CG, Collins-Underwood JR, Phillips LA, Loudin MG, Liu W, Zhang J, Ma J, Coustan-Smith E, Harvey RC, Willman CL, Mikhail FM, Meyer J, Carroll AJ, Williams RT, Cheng J, Heerema NA, Basso G, Pession A, Pui CH, Raimondi SC, Hunger SP, Downing JR, Carroll WL, Rabin KR. Rearrangement of CRLF2 in B-progenitor- and Down syndrome-associated acute lymphoblastic leukemia *Nat Genet* 2009 Nov;41(11):1243-6
- NCBI Resource Coordinators. Database resources of the National Center for Biotechnology Information *Nucleic Acids Res* 2016 Jan 4;44(D1):D7-19
- Niroula A, Vihinen M. Variation Interpretation Predictors: Principles, Types, Performance, and Choice *Hum Mutat* 2016 Jun;37(6):579-97
- Novo FJ, de Mendíbil IO, Vizmanos JL. TlCdb: a collection of gene-mapped translocation breakpoints in cancer *BMC Genomics* 2007 Jan 26;8:33
- Nowell PC, Hungerford DA. A minute Chromosome in Human Chronic Granulocytic Leukemia *Science* 1960 132:1497
- O'Leary NA, Wright MW, Brister JR, Ciufo S, Haddad D, McVeigh R, Rajput B, Robertse B, Smith-White B, Ako-Adjei D, Astashyn A, Badretdin A, Bao Y, Blinkova O, Brover V, Chetvernin V, Choi J, Cox E, Ermolaeva O, Farrell CM, Goldfarb T, Gupta T, Haft D, Hatcher E, Hlavina W, Joardar VS, Kodali VK, Li W, Maglott D, Masterson P, McGarvey KM, Murphy MR, O'Neill K, Pujar S, Rangwala SH, Rausch D, Riddick LD, Schoch C, Shkeda A, Storz SS, Sun H, Thibaud-Nissen F, Tolstoy I, Tully RE, Vatsan AR, Wallin C, Webb D, Wu W, Landrum MJ, Kimchi A, Tatusova T, DiCuccio M, Kitts P, Murphy TD, Pruitt KD. Reference sequence (RefSeq) database at NCBI: current status, taxonomic expansion, and functional annotation *Nucleic Acids Res* 2016 Jan 4;44(D1):D733-45
- Ohno S, Babonits M, Wiener F, Spira J, Klein G, Potter M. Nonrandom chromosome changes involving the Ig gene-carrying chromosomes 12 and 6 in pristane-induced mouse plasmacytomas *Cell* 1979 Dec;18(4):1001-7
- Oshimura M, Freeman AI, Sandberg AA. Chromosomes and causation of human cancer and leukemia XXVI. Binding studies in acute lymphoblastic leukemia (ALL)
- P&acute;aszczyca A, Nilsson J, Magnusson L, Brosjö O, Larsson O, Vult von Steyern F, Domanski HA, Lilljebjörn H, Fioretos T, Tayebwa J, Mandahl N, Nord KH, Mertens F. Fusions involving protein kinase C and membrane-associated proteins in benign fibrous histiocytoma *Int J Biochem Cell Biol* 2014 Aug;53:475-81
- Pagon RA. GeneTests: an online genetic information resource for health care providers *J Med Libr Assoc* 2006 Jul;94(3):343-8
- Pavlopoulou A, Spandidos DA, Michalopoulos I. Human cancer databases (review) *Oncol Rep* 2015 Jan;33(1):3-18
- Petryszak R, Keays M, Tang YA, Fonseca NA, Barrera E, Burdett T, Füllgrabe A, Fuentes AM, Jupp S, Koskinen S, Mannion O, Huerta L, Megy K, Snow C, Williams E, Barzine M, Hastings E, Weisser H, Wright J, Jaiswal P, Huber W, Choudhary J, Parkinson HE, Brazma A. Expression Atlas update--an integrated database of gene and protein expression in humans, animals and plants *Nucleic Acids Res* 2016 Jan 4;44(D1):D746-52
- Pinkel D, Albertson DG. Array comparative genomic hybridization and its applications in cancer *Nat Genet* 2005 Jun;37 Suppl:S11-7
- Pinkel D, Segraves R, Sudar D, Clark S, Poole I, Kowbel D, Collins C, Kuo WL, Chen C, Zhai Y, Dairkee SH, Ljung BM, Gray JW, Albertson DG. High resolution analysis of DNA copy number variation using comparative genomic hybridization to microarrays *Nat Genet* 1998 Oct;20(2):207-11
- Pundir S, Magrane M, Martin MJ, O'Donovan C; UniProt Consortium. Searching and Navigating UniProt Databases *Curr Protoc Bioinformatics* 2015 Jun 19;50:1
- Rath A, Olry A, Dhombres F, Brandt MM, Urbero B, Ayme S. Representation of rare diseases in health information systems: the Orphanet approach to serve a wide range of end users *Hum Mutat* 2012 May;33(5):803-8
- Redon R, Ishikawa S, Fitch KR, Feuk L, Perry GH, Andrews TD, Fiegler H, Shapero MH, Carson AR, Chen W, Cho EK, Dallaire S, Freeman JL, González JR, Gratacòs M, Huang J, Kalaitzopoulos D, Komura D, MacDonald JR, Marshall CR, Mei R, Montgomery L, Nishimura K, Okamura K, Shen F, Somerville MJ, Tchinda J, Valsesia A, Woodwark C, Yang F, Zhang J, Zerjal T, Zhang J, Armengol L, Conrad DF, Estivill X, Tyler-Smith C, Carter NP, Aburatani H, Lee C, Jones KW, Scherer SW, Hurles ME. Global variation in copy number in the human genome *Nature* 2006 Nov 23;444(7118):444-54
- Rickman DS, Pflueger D, Moss B, VanDoren VE, Chen CX, de la Taille A, Kuefer R, Tewari AK, Setlur SR, Demichelis F, Rubin MA. SLC45A3-ELK4 is a novel and frequent erythroblast transformation-specific fusion transcript in prostate cancer *Cancer Res* 2009 Apr 1;69(7):2734-8
- Rikova K, Guo A, Zeng Q, Possemato A, Yu J, Haack H, Nardone J, Lee K, Reeves C, Li Y, Hu Y, Tan Z, Stokes M, Sullivan L, Mitchell J, Wetzel R, Macneill J, Ren JM, Yuan J, Bakalarski CE, Villen J, Kornhauser JM, Smith B, Li D, Zhou X, Gygi SP, Gu TL, Polakiewicz RD, Rush J, Comb MJ. Global survey of phosphotyrosine signaling identifies oncogenic kinases in lung cancer *Cell* 2007 Dec 14;131(6):1190-203
- Roberts KG, Morin RD, Zhang J, Hirst M, Zhao Y, Su X, Chen SC, Payne-Turner D, Churchman ML, Harvey RC, Chen X, Kasap C, Yan C, Becksfort J, Finney RP, Teachey DT, Maude SL, Tse K, Moore R, Jones S, Mungall K, Birol I, Edmonson MN, Hu Y, Buetow KE, Chen IM, Carroll WL, Wei L, Ma J, Kleppe M, Levine RL, Garcia-Manero G, Larsen E, Shah NP, Devidas M, Reaman G, Smith M, Paugh SW, Evans WE, Grupp SA, Jeha S, Pui CH, Gerhard DS, Downing JR, Willman CL, Loh M, Hunger SP, Marra MA, Mullighan CG. Genetic alterations activating kinase and cytokine receptor signaling in high-risk acute



lymphoblastic leukemia *Cancer Cell* 2012 Aug 14;22(2):153-66

Rosenbloom KR, Armstrong J, Barber GP, Casper J, Clawson H, Diekhans M, Dreszer TR, Fujita PA, Guruvadoo L, Haussler M, Harte RA, Heitner S, Hickey G, Hinrichs AS, Hubley R, Karolchik D, Learned K, Lee BT, Li CH, Miga KH, Nguyen N, Paten B, Raney BJ, Smit AF, Speir ML, Zweig AS, Haussler D, Kuhn RM, Kent WJ. The UCSC Genome Browser database: 2015 update *Nucleic Acids Res* 2015 Jan;43(Database issue):D670-81

Rowley JD. Letter: A new consistent chromosomal abnormality in chronic myelogenous leukaemia identified by quinacrine fluorescence and Giemsa staining *Nature* 1973 Jun 1;243(5405):290-3

Rowley JD, Golomb HM, Dougherty C. 15/17 translocation, a consistent chromosomal change in acute promyelocytic leukaemia *Lancet* 1977 Mar 5;1(8010):549-50

Rubinstein WS, Maglott DR, Lee JM, Kattman BL, Malheiro AJ, Ovetsky M, Hem V, Gorelenkov V, Song G, Wallin C, Husain N, Chitipiralla S, Katz KS, Hoffman D, Jang W, Johnson M, Karmanov F, Ukrainchik A, Denisenko M, Fomous C, Hudson K, Ostell JM. The NIH genetic testing registry: a new, centralized database of genetic tests to enable access to comprehensive information and improve transparency *Nucleic Acids Res* 2013 Jan;41(Database issue):D925-35

Rutkowski P, Van Glabbeke M, Rankin CJ, Ruka W, Rubin BP, Debiec-Rychter M, Lazar A, Gelderblom H, Sciort R, Lopez-Terrada D, Hohenberger P, van Oosterom AT, Schuetze SM; European Organisation for Research and Treatment of Cancer Soft Tissue/Bone Sarcoma Group; Southwest Oncology Group. Imatinib mesylate in advanced dermatofibrosarcoma protuberans: pooled analysis of two phase II clinical trials *J Clin Oncol* 2010 Apr 1;28(10):1772-9

Santo EE, Ebus ME, Koster J, Schulte JH, Lakeman A, van Sluis P, Vermeulen J, Gisselsson D, Øra I, Lindner S, Buckley PG, Stallings RL, Vandesompele J, Eggert A, Caron HN, Versteeg R, Molenaar JJ. Oncogenic activation of FOXR1 by 11q23 intrachromosomal deletion-fusions in neuroblastoma *Oncogene* 2012 Mar 22;31(12):1571-81

Seidal T, Mark J, Hagmar B, Angervall L. Alveolar rhabdomyosarcoma: a cytogenetic and correlated cytological and histological study *Acta Pathol Microbiol Immunol Scand A* 1982 Sep;90(5):345-54

Shaffer LG, McGowen-Jordan J, Schmid M, editors. An International System for Human Cytogenetic Nomenclature 2013, Basel: S. Karger

Shaw AT, Hsu PP, Awad MM, Engelman JA. Tyrosine kinase gene rearrangements in epithelial malignancies *Nat Rev Cancer* 2013 Nov;13(11):772-87

Sigrist CJ, de Castro E, Cerutti L, Cuche BA, Hulo N, Bridge A, Bougueleret L, Xenarios I. New and continuing developments at PROSITE *Nucleic Acids Res* 2013 Jan;41(Database issue):D344-7

Simon MP, Pedetout F, Sirvent N, Grosgeorge J, Minoletti F, Coindre JM, Terrier-Lacombe MJ, Mandahl N, Craver RD, Blin N, Sozzi G, Turc-Carel C, O'Brien KP, Kedra D, Franssón I, Guilbaud C, Dumanski JP. Deregulation of the platelet-derived growth factor B-chain gene via fusion with collagen gene COL1A1 in dermatofibrosarcoma protuberans and giant-cell fibroblastoma *Nat Genet* 1997 Jan;15(1):95-8

Sinclair PB, Nacheva EP, Leversha M, Telford N, Chang J, Reid A, Bench A, Champion K, Huntly B, Green AR. Large deletions at the t(9;22) breakpoint are common and may identify a poor-prognosis subgroup of patients with chronic myeloid leukemia *Blood* 2000 Feb 1;95(3):738-43

Skotheim RI, Thomassen GO, Eken M, Lind GE, Micci F, Ribeiro FR, Cerveira N, Teixeira MR, Heim S, Rognes T, Lothe RA. A universal assay for detection of oncogenic fusion transcripts by oligo microarray analysis *Mol Cancer* 2009 Jan 19;8:5

Soda M, Choi YL, Enomoto M, Takada S, Yamashita Y, Ishikawa S, Fujiwara S, Watanabe H, Kurashina K, Hatanaka H, Bando M, Ohno S, Ishikawa Y, Aburatani H, Niki T, Sohara Y, Sugiyama Y, Mano H. Identification of the transforming EML4-ALK fusion gene in non-small-cell lung cancer *Nature* 2007 Aug 2;448(7153):561-6

Solinas-Toldo S, Lampel S, Stilgenbauer S, Nickolenko J, Benner A, Döhner H, Cremer T, Lichter P. Matrix-based comparative genomic hybridization: biochips to screen for genomic imbalances *Genes Chromosomes Cancer* 1997 Dec;20(4):399-407

Speicher MR, Carter NP. The new cytogenetics: blurring the boundaries with molecular biology *Nat Rev Genet* 2005 Oct;6(10):782-92

Steidl C, Shah SP, Woolcock BW, Rui L, Kawahara M, Farinha P, Johnson NA, Zhao Y, Telenius A, Neriah SB, McPherson A, Meissner B, Okoye UC, Diepstra A, van den Berg A, Sun M, Leung G, Jones SJ, Connors JM, Huntsman DG, Savage KJ, Rimsza LM, Horsman DE, Staudt LM, Steidl U, Marra MA, Gascoyne RD. MHC class II transactivator CIITA is a recurrent gene fusion partner in lymphoid cancers *Nature* 2011 Mar 17;471(7338):377-81

Stenman G, Sandros J, Dahlenfors R, Juberg-Ode M, Mark J. 6q- and loss of the Y chromosome--two common deviations in malignant human salivary gland tumors *Cancer Genet Cytogenet* 1986 Aug;22(4):283-93

Stephens PJ, McBride DJ, Lin ML, Varela I, Pleasance ED, Simpson JT, Stebbings LA, Leroy C, Edkins S, Mudie LJ, Greenman CD, Jia M, Latimer C, Teague JW, Lau KW, Burton J, Quail MA, Swerdlow H, Churcher C, Natrajan R, Sieuwerts AM, Martens JW, Silver DP, Langerød A, Russnes HE, Foekens JA, Reis-Filho JS, van't Veer L, Richardson AL, Børresen-Dale AL, Campbell PJ, Futreal PA, Stratton MR. Complex landscapes of somatic rearrangement in human breast cancer genomes *Nature* 2009 Dec 24;462(7276):1005-10

Stratton MR, Campbell PJ, Futreal PA. The cancer genome *Nature* 2009 Apr 9;458(7239):719-24

Sverre Heim and Felix Mitelman. *Cancer Cytogenetics: Chromosomal and Molecular Genetic Aberrations of Tumor Cells* 2015, Wiley-Blackwell, New-York

Swerdlow SH, Campo E, Pileri SA, Harris NL, Stein H, Siebert R, Advani R, Ghielmini M, Salles GA, Zelenetz AD, Jaffe ES. The 2016 revision of the World Health Organization (WHO) classification of lymphoid neoplasms *Blood* 2016 Mar 15

Tennessen JA, Bigham AW, O'Connor TD, Fu W, Kenny EE, Gravel S, McGee S, Do R, Liu X, Jun G, Kang HM, Jordan D, Leal SM, Gabriel S, Rieder MJ, Abecasis G, Altshuler D, Nickerson DA, Boerwinkle E, Sunyaev S, Bustamante CD, Bamshad MJ, Akey JM; Broad GO; Seattle GO; NHLBI Exome Sequencing Project. Evolution and functional impact of rare coding variation from deep sequencing of human exomes *Science* 2012 Jul 6;337(6090):64-9

Tomlins SA, Rhodes DR, Perner S, Dhanasekaran SM, Mehra R, Sun XW, Varambally S, Cao X, Tchinda J, Kuefer R, Lee C, Montie JE, Shah RB, Pienta KJ, Rubin MA, Chinnaiyan AM. Recurrent fusion of TMPRSS2 and ETS transcription factor genes in prostate cancer *Science* 2005 Oct 28;310(5748):644-8

- Turc-Carel C, Dal Cin P, Rao U, Karakousis C, Sandberg AA. Cytogenetic studies of adipose tissue tumors I A benign lipoma with reciprocal translocation t(3;12)(q28;q14)
- UniProt Consortium. UniProt: a hub for protein information Nucleic Acids Res 2015 Jan;43(Database issue):D204-12
- Urakami K, Shimoda Y, Ohshima K, Nagashima T, Serizawa M, Tanabe T, Saito J, Usui T, Watanabe Y, Naruoka A, Ohnami S, Ohnami S, Mochizuki T, Kusuvara M, Yamaguchi K. Next generation sequencing approach for detecting 491 fusion genes from human cancer Biomed Res 2016;37(1):51-62
- Van Den Berghe H, Gosseye CP, Englebienne V, Cornu G, Sokal G. Variant translocation in Burkitt lymphoma Cancer Genetics and Cytogenetics 1960; 1; 9-14
- Van Vlierberghe P, van Grotel M, Tchinda J, Lee C, Beverloo HB, van der Spek PJ, Stubbs A, Cools J, Nagata K, Fornerod M, Buijs-Gladdines J, Horstmann M, van Wering ER, Soulier J, Pieters R, Meijerink JP. The recurrent SET-NUP214 fusion as a new HOXA activation mechanism in pediatric T-cell acute lymphoblastic leukemia Blood 2008 May 1;111(9):4668-80
- Wang L, Motoi T, Khanin R, Olshen A, Mertens F, Bridge J, Dal Cin P, Antonescu CR, Singer S, Hameed M, Bovee JV, Hogendoorn PC, Socci N, Ladanyi M. Identification of a novel, recurrent HEY1-NCOA2 fusion in mesenchymal chondrosarcoma based on a genome-wide screen of exon-level expression data Genes Chromosomes Cancer 2012 Feb;51(2):127-39
- Wang Y, Wu N, Liu J, Wu Z, Dong D. FusionCancer: a database of cancer fusion genes derived from RNA-seq data Diagn Pathol 2015 Jul 28;10:131
- Watanabe M, Serizawa M, Sawada T, Takeda K, Takahashi T, Yamamoto N, Koizumi F, Koh Y. A novel flow cytometry-based cell capture platform for the detection, capture and molecular characterization of rare tumor cells in blood J Transl Med 2014 May 23;12:143
- Welch JS, Westervelt P, Ding L, Larson DE, Kline JM, Kulkarni S, Wallis J, Chen K, Payton JE, Fulton RS, Veizer J, Schmidt H, Vickery TL, Heath S, Watson MA, Tomasson MH, Link DC, Graubert TA, DiPersio JF, Mardis ER, Ley TJ, Wilson RK. Use of whole-genome sequencing to diagnose a cryptic fusion oncogene JAMA 2011 Apr 20;305(15):1577-84
- West RB, Rubin BP, Miller MA, Subramanian S, Kaygusuz G, Montgomery K, Zhu S, Marinelli RJ, De Luca A, Downs-Kelly E, Goldblum JR, Corless CL, Brown PO, Gilks CB, Nielsen TO, Huntsman D, van de Rijn M. A landscape effect in tenosynovial giant-cell tumor from activation of CSF1 expression by a translocation in a minority of tumor cells Proc Natl Acad Sci U S A 2006 Jan 17;103(3):690-5
- Wildschutte JH, Williams ZH, Montesin M, Subramanian RP, Kidd JM, Coffin JM. Discovery of unfixed endogenous retrovirus insertions in diverse human populations Proc Natl Acad Sci U S A 2016 Apr 19;113(16):E2326-34
- Wu C, Jin X, Tsung G, Afrasiabi C, Su AI. BioGPS: building your own mash-up of gene annotations and expression profiles Nucleic Acids Res 2016 Jan 4;44(D1):D313-6
- Wu J, Wu M, Li L, Liu Z, Zeng W, Jiang R. dbWGF: a database and web server of human whole-genome single nucleotide variants and their functional predictions Database (Oxford) 2016 Mar 17;2016
- Wu TJ, Shamsaddini A, Pan Y, Smith K, Crichton DJ, Simonyan V, Mazumder R. A framework for organizing cancer-related variations from existing databases, publications and NGS data using a High-performance Integrated Virtual Environment (HIVE) Database (Oxford) 2014 Mar 25;2014:bau022
- Yang Y, Dong X, Xie B, Ding N, Chen J, Li Y, Zhang Q, Qu H, Fang X. Databases and web tools for cancer genomics study Genomics Proteomics Bioinformatics 2015 Feb;13(1):46-50
- Yates A, Akanni W, Amode MR, Barrell D, Billis K, Carvalho-Silva D, Cummins C, Clapham P, Fitzgerald S, Gil L, Girón CG, Gordon L, Hourlier T, Hunt SE, Janacek SH, Johnson N, Juettemann T, Keenan S, Lavidas I, Martin FJ, Maurel T, McLaren W, Murphy DN, Nag R, Nuhn M, Parker A, Patricio M, Pignatelli M, Rahtz M, Riat HS, Sheppard D, Taylor K, Thormann A, Vullo A, Wilder SP, Zadissa A, Birney E, Harrow J, Muffato M, Perry E, Ruffier M, Spudich G, Trevanion SJ, Cunningham F, Aken BL, Zerbino DR, Flicek P. Ensembl 2016 Nucleic Acids Res 2016 Jan 4;44(D1):D710-6
- Yoshihara K, Wang Q, Torres-Garcia W, Zheng S, Vegesna R, Kim H, Verhaak RG. The landscape and therapeutic relevance of cancer-associated transcript fusions Oncogene 2015 Sep 10;34(37):4845-54
- Yu KH, Ricigliano M, Hidalgo M, Abou-Alfa GK, Lowery MA, Saltz LB, Crotty JF, Gary K, Cooper B, Lapidus R, Sadowska M, O'Reilly EM. Pharmacogenomic modeling of circulating tumor and invasive cells for prediction of chemotherapy response and resistance in pancreatic cancer Clin Cancer Res 2014 Oct 15;20(20):5281-9
- Yue P, Moul J. Identification and analysis of deleterious human SNPs J Mol Biol 2006 Mar 10;356(5):1263-74
- Zech L, Haglund U, Nilsson K, Klein G. Characteristic chromosomal abnormalities in biopsies and lymphoid-cell lines from patients with Burkitt and non-Burkitt lymphomas Int J Cancer 1976 Jan 15;17(1):47-56
- Zhang J, Baran J, Cros A, Guberman JM, Haider S, Hsu J, Liang Y, Rivkin E, Wang J, Whitty B, Wong-Erasmus M, Yao L, Kasprzyk A. International Cancer Genome Consortium Data Portal--a one-stop shop for cancer genomics data Database (Oxford) 2011 Sep 19;2011:bar026
- de Jong B, Molenaar IM, Leeuw JA, Idenberg VJ, Oosterhuis JW. Cytogenetics of a renal adenocarcinoma in a 2-year-old child Cancer Genet Cytogenet 1986 Mar 15;21(2):165-9
- de Klein A, van Kessel AG, Grosveld G, Bartram CR, Hagemeijer A, Bootsma D, Spurr NK, Heisterkamp N, Groffen J, Stephenson JR. A cellular oncogene is translocated to the Philadelphia chromosome in chronic myelocytic leukaemia Nature 1982 Dec 23;300(5894):765-7

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